Congenital renal abnormalities in fetuses: a study in a teaching institute

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Abstract

Introduction: The term renal malformations include a number of development aberrations which include structural and developmental anomalies. Renal agenesis, Renal dysplasia, hypoplasia and polycystic kidney disease are some of the renal anomalies.

Materials and Method: The approximate age, sex and crown rump length were recorded for the 50 foetuses included in the study. The embalmed fetuses were dissected as per protocol and anomalies were recorded. The size, shape and the kidneys, the arrangement of the attached structures such as the hilum, ureter, bladder abdominal aorta and the inferior vena cava were noted and recorded.

Results: out of the 50 foetuses 29 were males and 21 were females. 2 of the fetuses had abnormal kidneys, out of which, one was male with Right Renal agenesis and the other was female with congenital polycystic kidney. The surface of both the kidneys of the male fetus showed cysts which looked like gas bubbles, thereby showing the presence of congenital polycystic kidney. There was agenesis observed in the right kidney and ureter in the female fetus while the left kidney was normal.

Conclusion: Although the renal anomalies are relatively uncommon, it is better for early detection so that corrective measures can be taken either pre or postnatally.

Keywords: Renal abnormality, Renal agenesis, Congenital polycystic renal disease, Fetus

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Introduction

Congenital abnormalities of the urinary tract are most commonly identified sonographically. Their incidence is said to occur in every 1-4 pregnancies,(1) with a representation of 15-20% of all prenatally diagnosed congenital anomalies. Obstructive uropathy accounts for majority of the cases.(2) With the advent of ultra sound, these anomalies are being detected very early in life and many of the newborn are being treated within the first five years of life, although, there are quite a few fatalities in utero also.(3)

The urinary tract development take place in the third week of intrauterine life form the intermediate mesoderm and form the urogenital sinus (bladder and the urethra), while the intermediate mesoderm form the kidneys and the ureters. During this development, the renal system pass through the pro and mesonephros and the metanephros stage before forming the definitive kidney.(4) From about 9-12 weeks of pregnancy, the fetal kidneys and adrenal glands are well developed to be visualized on either sides of the lumbar spine.

The term renal malformations include a number of development aberrations which include structural and developmental anomalies. Renal agenesis is one such example where the kidney is totally absent.(5,6) Renal dysplasia occurs when there are undifferentiated and metaplastic cells within the organs. These many be tiny as in the case of renal aplasia or can distend the abdomen as in multicystic dysplastic kidney. A hypoplastic kidney is small with very few nephrons than normal, which may be greatly enlarged in oligomeganephronia.(7)

Some other type of anomalies is the polycystic kidney disease, which include autosomal recessive polycystic kidneys and the disorder resulting from genetic mutations.(8,11)

In an analysis by the Northern Congenital Abnormality Survey (NorCAS), 19.6% of the pregnancies, in which a congenital anomaly was identified failed to produce a living child. Therefore, this study was conducted to identify the renal anomalies present in the fetuses which do not produce a living child.

Materials and Method

This study was conducted by the Department of Anatomy at Mediciti institute of medical sciences during the period of two years. A total of 50 dead newborn were dissected and thoroughly verified. The information was collected from the autopsies conducted by the Forensic department of our hospital.

The still born were brought from the maternity hospitals after the proper paperwork, and were kept in 10% formalin solution. The approximate age, sex and crown rump length were recorded for each foetus. All the foetuses were properly numbered and were injecting with preservative liquids by body injectors and storing in the tank solution. They were dissected after 3 or 4 days and if no anomaly was found, they were discarded.

The dissection was performed with a midline incision from supra-sternal notch to symphysis pubis and two transverse incision form the umbilicus laterally as far as possible to expose the abdominal and the thoracic cavities completely. The position of the
suprarenal gland and the upper poles of the kidneys were noted and recorded. The size, shape and the kidneys were also recorded. The arrangement of the attached structures such as the hilum, ureter, bladder abdominal aorta and the inferior vena cava were noted and recorded.

Results

Out of the 50 foetuses 29 were males and 21 were females. 2 of the fetuses had abnormal kidneys, out of which, one was male with Right Renal agenesis and the other was female with congenital polycystic kidney. The male was of 28 gestation weeks with a crown rump length of 27cms and head circumference of 15 cms. All the external features were normal. Internally, the gastrointestinal area i.e. the stomach to the sigmoid colon was found to be normal and removed. The right and left kidneys were slightly larger than a normal sized kidney of the same gestational age and measured 6.5 x 3 x 1cm. The surface of both the kidneys showed cysts which looked like gas bubbles, thereby showing the presence of congenital polycystic kidney (Fig. 1).

Fig. 1: Congenital polycystic kidney

The rest of the features such as position of the suprarenal glands, structures at hilum, renal vein, renal artery, and the ureter were normal. The right ureter was dilated but normal in size. There was no stenosis, no tortuosity and the renal pelvis was uniformly dilated. Similar was the case with the left ureter.

In the second fetus which was a female, the gestational age was 26 weeks with the crown rump being 25 cms. The rest of the external features were normal.

There was agenesis observed in the right kidney and ureter. The right supra renal gland was very small in size, though the left kidney, ureter and the supra renal gland were normal (Fig. 2).

Discussion

Though very few studies have been conducted on the survey of congenital malformations in the nonliving fetuses, there are a few in the living fetuses. We have attempted to compare the cases from both live and nonliving fetus studies.

In the present study, congenital anomalies of the kidneys was found in 4% of the fetuses. An abnormality in the development of kidney and ureter was found in 9.6% by Dees et al. An incidence of renal abnormalities was found in 1 in 27 individuals (3.7%) by Smith et al while Culp et al found in 3.1% of the cases. In a more recent study by Scott, 57.7% of the deaths of the fetuses was solely due to renal anomalies.

With more recent studies it was unable to compare due to procedural reasons. For instance, some studies do not register cases delivered before 20 weeks of gestation and include only renal agenesis and cystic diseases for urinary tract anomalies. In another instance, 45% of the pregnancies were terminated but few major anomalies went undetected.

Of all the anomalies, unilateral agenesis occurs more commonly, though bilateral agenesis has a profound effect on the patient. Very few cases have been cited for bilateral agenesis with most of them being cited by Potter in 1965.

We have not come across a bilateral agenesis but did come across a unilateral agenesis in one female fetus. In a study by Davidson et al, a 0.28% incidence was observed of bilateral anomaly with a significantly higher rate in males rather than females.

Renal agenesis is said to occur when there is an absence of the metanephrogenic blastema, or absence of the ureteric bud. This disorder may also be associated with genital tract abnormality, skeletal abnormality, anorectal malformation and cryptorchidism. There was no adrenal gland in the agenesis patient in our study. It is estimated that in 8-17% of the cases, there will be no adrenal gland for such cases.

We had encountered a case of congenital polycystic kidney in one male fetus. In a study by Scott, 78 out of 323 patients with renal anomaly had polycystic renal disease.


Autosomal recessive
polycystic kidney disease has a spectrum of severity, with most of the severe forms appearing earliest in life and being bilateral. Usually those diagnosed early in life are usually recessive type.20

It is estimated that the incidence of autosomal recessive polycystic kidney disease is 1 in 20,000 live births.21 It is more frequently said to occur due to a mutation in the PKHD-1 gene, but additional genes are also known to play a role. A heterozygous carrier state is reported in 1 in 70.22 It is characterized by the dilatation of tubules especially in the medulla, with the outer cortex being spared.19 Severe affected fetuses if born are born with oligohydramnios, Potter’s face. Many of them will develop respiratory insufficiently. In survivors, approximately 40% have severe hepatic and renal disease.14

This disease basically results in bilateral symmetric microcystic disease occurring in the distal convoluted tubules and collecting ducts,18 maybe associated with Caroli’s disease23 or congenital hepatic fibrosis.24,25 Secondary to the liver fibrosis, they may develop portal hypertension, esophageal varices, hemorrhoids, upper gastrointestinal bleeding, splenomegaly and hypersplenism.

The limitation in this study was the less number of specimens for study.

Conclusion

The number of dead fetuses due to renal anomalies have not improved in the past few years probably due to early detection and medical termination of pregnancy rather than post natal deaths. Many of these anomalies are hereditary and due to the advances in prenatal diagnosis, advent of fetal surgeries and kidney transplants, there has been an improvement in the prognosis although still the numbers are not statistically significant. Proper diagnosis of the genetic abnormalities are possible in today field of medicine which can improve and prevent further fetal anomalies.

References